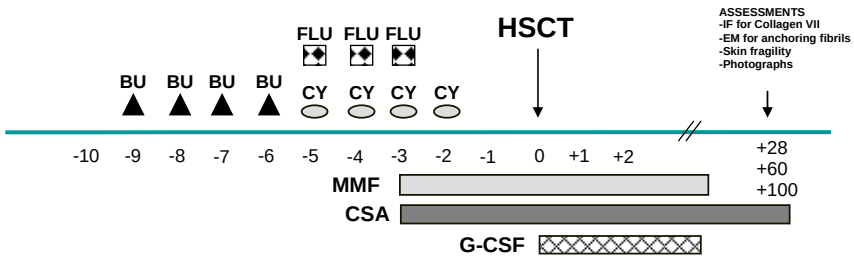
Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

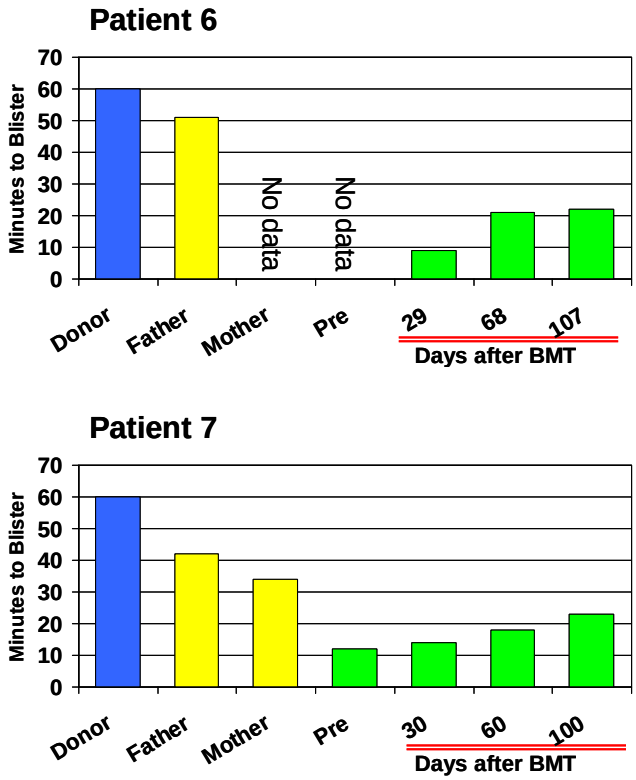
Supplement to: Wagner JE, Ishida-Yamamoto A, McGrath JA, et al. Bone marrow transplantation for recessive dystrophic epidermolysis bullosa. N Engl J Med 2010;363:629-39.

Figure 1 Treatment Schema



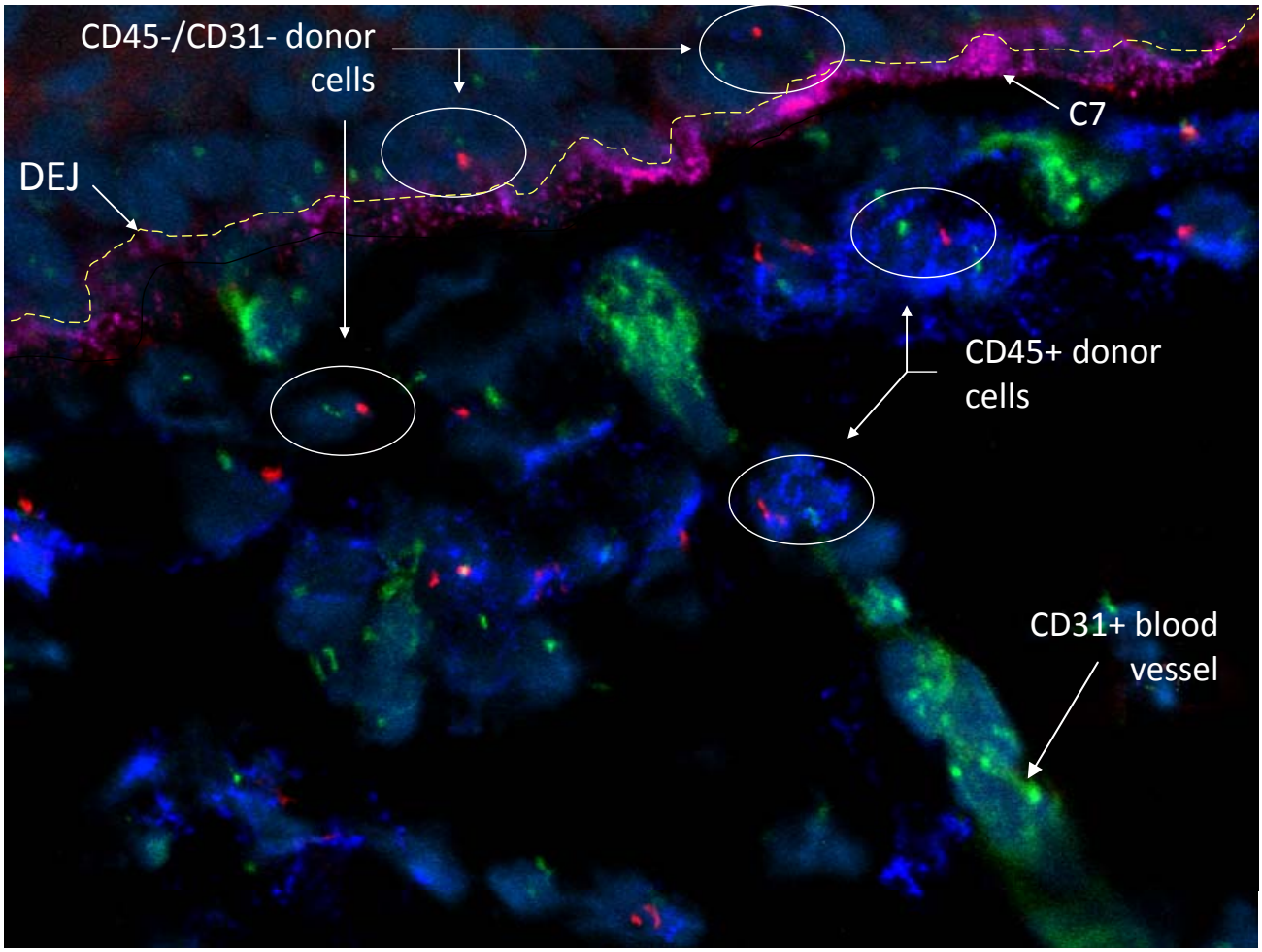
Prior to BM infusion, all patients received immunomyeloablation consisting of busulfan, cyclophosphamide and fludarabine. On day 0, hematopoietic stem cells from BM or umbilical cord blood were infused without filtering or processing. Immunoprophylaxis for acute graft-versus-host disease consisted of cyclosporine A for 6 months and mycophenolate mofetil for 1 month after BM infusion.

Figure 2 Blister Test



Two 3-millimeter diameter patches of skin on the upper and lower extremity were exposed to continuous negative pressure (12 inches of mercury). Time to blister formation was determined with a stop watch. The average of the two measurements pretransplant and for the specified time points after transplant are shown for patients 6 and 7. Measurements of skin resistance are also reported for the patient's donor, mother and father. Results indicate increased resistance after marrow transplant for patients 6 and 7.

Figure 3 Analysis of Donor Cells in Recipient Skin.



Combined immunofluorescence and fluorescence *in situ* hybridization (FISH) was used to identify the lineage and donor (XY) or recipient (XX) origin of cells in the skin of Patient 5. Donor cells (circled) expressing pan-hematopoietic CD45 antigen (dark blue) were identified in the papillary dermis below the DEJ. Donor cells not expressing pan-hematopoietic or endothelial antigens CD45 and CD31 were identified in the epidermis above the DEJ as well as the papillary dermis. A linear band of C7 was detected with anti-C7 antibody (fuchsia). Donor cells were found predominantly outside blood vessels (labeled green with anti-CD31 antibody). FISH probes: Y (red), X (green).